

MSUD (Maple Syrup Urine Disease) – Increased Leucine Screening
Differential Diagnosis: Maple Syrup Urine Disease (MSUD)
 Oklahoma State Department of Health Newborn Screening Program

MSUD Screen
Out-of-Range (abnormal)
MEDICAL EMERGENCY

NO

YES

Not consistent with MSUD.
 No further action needed.

Primary Analyte

Leucine $\geq 355 \mu\text{mol/L}$

Table 1. In-range MSUD Screen Results¹:

Primary Analyte:
 Leucine $< 355 \mu\text{mol/L}$

Secondary Analyte²:
 Valine $< 330 \mu\text{mol/L}$

¹These values are utilized for newborns less than 60 days old.

²Elevations of the secondary analytes are reported as “not consistent with MSUD” if leucine is in-range.

From time of report, the contacted provider will:

1. Contact family within one hour of notification.
2. Immediately consult with metabolic specialist.
3. In consultation with the metabolic specialist, arrange clinical evaluation before COB utilizing the Emergency Management Protocol (initial assessment can be done by provider or metabolic specialist).
4. Refer for immediate evaluation and diagnostic work-up in consultation with Metabolic Specialist. Evaluation by metabolic specialist must occur within 8 hours.

Appointment with metabolic specialist for diagnostic testing (testing must be coordinated by the specialist or newborn screening program):

1. Plasma amino acid (plasma)
2. Urine Organic Acids
3. Other lab and/or DNA may be indicated

Diagnostic Testing Inconclusive:
 Monitoring and medical management as advised by metabolic specialist.

Diagnostic Testing Consistent with: Refer to metabolic specialist for medical management.

Diagnostic Testing Within Normal Limits: Not consistent with MSUD. No further follow-up indicated.

From the time the screen is reported to the provider, the Metabolic Nurse Specialist will monitor follow-up by:

1. Confirming the provider contacts family within one hour of notification.
2. Facilitating and confirming a clinical evaluation by a provider or metabolic specialist is achieved before COB.
3. Facilitating and confirming infant presents for a diagnostic workup with a metabolic specialist within 24 hours.
4. Coordinating collection and processing of diagnostic tests and communicating test results to provider and short-term follow-up program (STFU).
5. Communicating with STFU if the above timelines are not met.